References

[1] Moser HW, Mahmood A, Raymond GV. X-linked adrenoleukodystrophy. Nat Clin Pract Neurol. 3(3)(March 2007): 140-51.

[2] Moser HW, Moser AB, Frayer KK, Chen W, Schulman JD, O'Neill BP, et al. Adrenoleukodystrophy: increased plasma content of saturated very long chain fatty acids. Neurology. 31(10)(October 1981): 1241-9.

[3] Mosser J, Douar AM, Sarde CO, Kioschis P, Feil R, Moser H, et al. Putative X-linked adrenoleukodystrophy gene shares unexpected homology with ABC transporters. Nature. 361(6414)(February 1993): 726-30.

[4] Liu YT, Lin KH, Soong BW, Liao KK, Lin KP. A novel ABCD 1 gene mutation in a Chinese-Taiwanese patient with adrenomyeloneuropathy. Pediatr Neurol. 36(5)(May 2007): 348-50.

[5] Kroos MA, Mullaart RA, Van Vliet L, Pomponio RJ, Amartino H, Kolodny EH, et al. p.[G576S; E689K]: pathogenic combination or polymorphism in Pompe disease? Eur J Hum Genet. 16(8)(August 2008): 875-9.

[6] Pittis MG, Donnarumma M, Montalvo AL, Dominissini S, Kroos M, Rosano C, et al. Molecular and functional characterization of eight novel GAA mutations in Italian infants with Pompe disease. Hum Mutat. 29(6)(June 2008): E27-36.

[7] Minegishi Y, Karasuyama H. Genetic origins of hyper-IgE syndrome. Curr Allergy Asthma Rep. 8(5)(September 2008): 386-91.

[8] Grimbacher B, Holland SM, Gallin JI, Greenberg F, Hill SC, Malech HL, et al. Hyper-IgE syndrome with recurrent infections--an autosomal dominant multisystem disorder. N Engl J Med. 340(9)(March 1999): 692-702.

[9] Freeman AF, Kleiner DE, Nadiminti H, Davis J, Quezado M, Anderson V, et al. Causes of death in hyper-IgE syndrome. J Allergy Clin Immunol. 119(5) (May 2007): 1234-40.

[10] Holland SM, DeLeo FR, Elloumi HZ, Hsu AP, Uzel G, Brodsky N, et al. STAT 3 mutations in the hyper-IgE syndrome. N Engl J Med. 357(16)(October 2007): 1608-19.

[11] Minegishi Y, Saito M, Tsuchiya S, Tsuge I, Takada H, Hara T, et al. Dominant-negative mutations in the DNA-binding domain of STAT3 cause hyper-IgE syndrome. Nature. 448(7157)(August 2007): 1058-62.

[12] Renner ED, Puck JM, Holland SM, Schmitt M, Weiss M, Frosch M, et al. Autosomal recessive hyperimmunoglobulin E syndrome: a distinct disease entity. J Pediatr. 144(1)(January 2004): 93-9.

[13] Minegishi Y, Saito M, Morio T, Watanabe K, Agematsu K, Tsuchiya S, et al. Human tyrosine kinase 2 deficiency reveals its requisite roles in multiple cytokine signals involved in innate and acquired immunity. Immunity. 25(5)(November 2006): 745-55.

[14] Levy DE, Loomis CA. STAT3 signaling and the hyper-IgE syndrome. N Engl J Med. 357(16)(October 2007): 1655-8.

[15] Jiao H, Toth B, Erdos M, Fransson I, Rakoczi E, Balogh I, et al. Novel and recurrent STAT3 mutations in hyper-IgE syndrome patients from different ethnic groups. Mol Immunol. 46(1)(November 2008): 202-6.

[16] Heinritz W, Shou L, Moschik A, Froster UG. The human TBX5 gene mutation database. Hum Mutat. 26(4)(October 2005): 397.

[17] Holt M, Oram S. Familial heart disease with skeletal malformations. Br Heart J. 22(April 1960): 236-42.

[18] Basson CT, Bachinsky DR, Lin RC, Levi T, Elkins JA, Soults J, et al. Mutations in human TBX5 [corrected] cause limb and cardiac malformation in Holt-Oram syndrome. Nat Genet. 15(1)(January 1997): 30-5.

[19] Bruneau BG, Nemer G, Schmitt JP, Charron F, Robitaille L, Caron S, et al. A murine model of Holt-Oram syndrome defines roles of the T-box transcription factor Tbx5 in cardiogenesis and disease. Cell. 106(6)(September 2001): 709-21.

[20] Fan C, Liu M, Wang Q. Functional analysis of TBX5 missense mutations associated with Holt-Oram syndrome. J Biol Chem. 278(10)(March 2003): 8780-5.

[21] Becker K, Beales PL, Calver DM, Matthijs G, Mohammed SN. Okihiro syndrome and acro-renal-ocular syndrome: clinical overlap, expansion of the phenotype, and absence of PAX2 mutations in two new families. J Med Genet. 39(1)(January 2002): 68-71.

[22] Kohlhase J, Schubert L, Liebers M, Rauch A, Becker K, Mohammed SN, et al. Mutations at the SALL4 locus on chromosome 20 result in a range of clinically overlapping phenotypes, including Okihiro syndrome, Holt-Oram syndrome, acro-renal-ocular syndrome, and patients previously reported to represent thalidomide embryopathy. J Med Genet. 40(7)(July 2003): 473-8.

[23] Hochberg MC. Systemic lupus erythematosus. Rheum Dis Clin North Am. 16(3)(August 1990): 617-39.

[24] Santiago-Raber ML, Laporte C, Reininger L, Izui S. Genetic basis of murine lupus. Autoimmun Rev. 3(1)(January 2004): 33-9.

[25] Weintraub JP, Godfrey V, Wolthusen PA, Cheek RL, Eisenberg RA, Cohen PL. Immunological and pathological consequences of mutations in both Fas and Fas ligand. Cell Immunol. 186(1)(May 1998): 8-17.

[26] Mysler E, Bini P, Drappa J, Ramos P, Friedman SM, Krammer PH, et al. The apoptosis-1/Fas protein in human systemic lupus erythematosus. J Clin Invest. 93(3)(March 1994): 1029-34.

[27] Costenbader KH, Feskanich D, Stampfer MJ, Karlson EW. Reproductive and menopausal factors and risk of systemic lupus erythematosus in women. Arthritis Rheum. 56(4)(April 2007): 1251-62.

[28] Kishnani PS, Steiner RD, Bali D, Berger K, Byrne BJ, Case LE, et al. Pompe disease diagnosis and management guideline. Genet Med. 8(5)(May 2006): 267-88.

[29] Levy DE, Lee CK. What does Stat3 do? J Clin Invest. 109(9)(May 2002): 1143-8.

[30] Encinas JA, Kuchroo VK. Mapping and identification of autoimmunity genes. Curr Opin Immunol. 12(6)(December 2000): 691-7.

[31] Han B, Bojalil R, Amezcua-Guerra LM, Springall R, Valderrama-Carvajal H, Wu J, et al. DcR3 as a diagnostic parameter and risk factor for systemic lupus erythematosus. Int Immunol. 20(8)(August 2008): 1067-75.

[32] Han B, Moore PA, Wu J, Luo H. Overexpression of human decoy receptor 3 in mice results in a systemic lupus erythematosus-like syndrome. Arthritis Rheum. 56(11)(November 2007): 3748-58.

[33] Lee CS, Hu CY, Tsai HF, Wu CS, Hsieh SL, Liu LC, et al. Elevated serum decoy receptor 3 with enhanced T cell activation in systemic lupus erythematosus. Clin Exp Immunol. 151(3)(March 2008): 383-90.

APPENDICES

APPENDIX A

BUFFERS AND REAGENT

1. **10X Tris borate buffer (10X TBE buffer)**

Tris – base	100	g
Boric acid	55	g
0.5 M EDTA (pH 8.0)	40	ml

Adjust volume to 1,000 ml with distilled water. The solution was mixed and stored at room temperature.

2. **6X loading dye**

Bromphenol blue	0.25	g
Xylene cyanol	0.25	g
Glycerol	50	ml
1M Tris (pH 8.0)	1	ml
Distilled water until	100	ml

Mix and store at 4^0C

3. **2% Agarose gel (w/v)**

Agarose	2.0	g
1X TBE	100	ml

Dissolve by heating in microwave oven and occasionally mix until no granules of agarose are visible.

4. **Ethidium bromide**

Ethidium bromide	10	mg
Distilled water	1	ml

Mix the solution and store at 4^0C

5. **Phosphate-Buffered Saline (PBS)**

Solution A

NaCl	8.0	g
KCl	0.2	g
$CaCl_2.2H_2O$	0.132	g
$MgCl_2.6H_2O$	0.1	g
Distilled water	800	ml

Solution B

Na_2HPO_4	1.15 g
KH2PO4	0.2 g
Distilled water	800 ml

Dissolve each solution in demineralized water. Autoclave solutions A and B separately at 15 pounds for 15 minutes. Mix A and B when cold: stir slowly; final pH 7.0 and store at 4^0C.

6. **100 bp ladder**

100 bp ladder stock	30 µl
TBE buffer	30 µl
1X loading dye	30 µl

Mix the solution and store at 4^0C

7. **1k bp ladder**

1k bp ladder stock	30 µl
TBE buffer	30 µl
1X loading dye	30 µl

Mix the solution and store at 4^0C

APPENDIX B

SAMPLE SIZE

Sample size (two independent groups) for ELIZA experiment

From previous report [31]

Patients (n_1) = 90	controls (n_2) = 123
mean ($\bar{X}_1$) = 32	mean ($\bar{X}_2$) = 5
SD (S_1) = $\pm$ 32	SD (S_2) = $\pm$ 12

Calculation

α = 0.05

β = 0.10

$Z_{\alpha/2} = Z_{0.05/2} = 1.96$ (two tails)

$Z_{\beta} = Z_{0.10} = 1.28$

$$\text{n/group} = 2(Z_{\alpha/2} + Z_{\beta})^2 \sigma^2 / (\bar{X}_1 - \bar{X}_2)^2$$

$\bar{X}_1$ = mean of patients

$\bar{X}_2$ = mean of controls

σ^2 = Pooled variance

$$= \frac{(n_1-1)S_1^2 + (n_2-1)S_2^2}{n_1+n_2-2}$$

$$= \frac{(90-1)32^2 + (123-1)12^2}{90+123-2}$$

$$= 515.185$$

$$\text{n/group} = \frac{2(1.96+1.28)\times 515.185}{(32-5)^2}$$

$$= 14.8$$

APPENDIX C

CRITERIA FOR SELECTION OF CONTROLS & RESULT OF ELISA

Controls of serum DcR3 levels using ELISA

In this study, serum from healthy children could not be obtained. Therefore, we used control serum from patients who met the following criteria.

Inclusion criteria for selection of the unaffected controls

1. Age

   ≤ 20 years old

2. Patients without the following disorders;

   2.1 Myeloma

   2.2 Nasopharyngeal carcinoma

   2.3 Pituitary adenoma

   2.4 Hepatocellular carcinoma

   2.5 Crohn's disease

   2.6 Pancreatic adenocarcinoma

   2.7 Gastric cancer

   2.8 Renal cancer

   2.9 Epithelial ovarian cancer

   2.10 B cell lymphoma

   2.11 Laryngeal carcinoma

   2.12 Ovarian cancer

   2.13 Glioma

   2.14 Colorectal cancer

   2.15 Gastrointestinal tract tumor

   2.16 Silicosis

   2.17 Leukemia

   2.18 Kidney transplant

3. The patients with mild illnesses

4. The patients/parents signed the consent form.

Table17. Evaluation of serum DcR3 (pg/μl) in SLE patients by ELISA

Plate ID: 7 **Biotrak II Reader** Results

Test : DCR3450/620
Measurement Date : 03.08.09 17:12
Measurement Filters : 450/620 nm

Legend: Layout / Absorbance / Concentration / Thresholds
Concentration range: 51.4526 pg/mL .. 7579.6 pg/mL
Thresholds not available.

	1	2	3	4	5	6	7	8	9	10	11	12
A	BK 0.003	ST4 0.191 608.9	SM1 -0.015 1852	SM9 -0.005 1865	SM17 -0.00? 1859	SM25 -0.004 1869	SM33 -0.005 1865	SM41 0.002 69.96	SM49 -0.010 1849	SM57 0.050 289.5	SM65 -0.008 1855	SM73 -0.010 1859
B	BK -0.002	ST4 0.170 569	SM2 -0.021 1913	SM10 -0.010 1849	SM18 0.020 185	SM26 -0.016 1829	SM34 -0.011 1845	SM42 -0.008 1855	SM50 0.393 961.7	SM58 0.036 245.4	SM66 -0.012 1842	SM74 0.038 385.6
C	ST1 2.900 4720	ST5 0.075 352.9	SM3 0.070 345.1	SM11 -0.020 1816	SM19 0.020 185	SM27 -0.021 1812	SM35 0.004 91.96	SM43 0.012 146.6	SM51 0.021 189.3	SM59 0.029 220.7	SM67 -0.004 1869	SM75 -0.004 1869
D	ST1 2.962 4649	ST5 0.048 283.9	SM4 0.001 53.95	SM12 -0.008 1855	SM20 -0.01? 1845	SM28 -0.021 1813	SM36 -0.016 1829	SM44 -0.011 1845	SM52 0.127 479.3	SM60 0.018 176.2	SM68 0.009 129.2	SM76 0.004 91.96
E	ST2 1.507 2347	ST6 0.018 176.2	SM5 -0.019 1819	SM13 -0.019 1819	SM21 -0.020 1791	SM29 -0.018 1823	SM37 -0.005 1865	SM45 -0.026 1797	SM53 0.608 1299	SM61 0.035 241.9	SM69 0.184 595.4	SM77 1.540 3257
F	ST2 1.744 2968	ST6 0.014 157.06	SM6 0.016 166.9	SM14 -0.023 1806	SM22 -0.022 1810	SM30 -0.019 1819	SM38 -0.025 1800	SM46 -0.003 1872	SM54 -0.013 1839	SM62 0.024 201.6	SM70 0.231 683.8	SM78 0.051 292.4
G	ST3 0.472 1088	ST7 0.001 53.95	SM7 0.284 778.2	SM15 0.246 711.01	SM23 -0.011 1845	SM31 -0.018 1823	SM39 -0.021 1813	SM47 0.051 292.4	SM55 -0.025 1800	SM63 0.083 378.2	SM71 0.138 502.7	SM79 0.286 781.8
H	ST3 0.530 1256	ST7 0.006 [illegible]	SM8 0.039 255.3	SM16 -0.006 1862	SM24 0.008 414.3	SM32 0.002 69.96	SM40 -0.003 1872	SM48 -0.017 1826	SM56 0.033 235.05	SM64 0.001 53.95	SM72 0.019 180.6	SM80 0.131 487.9

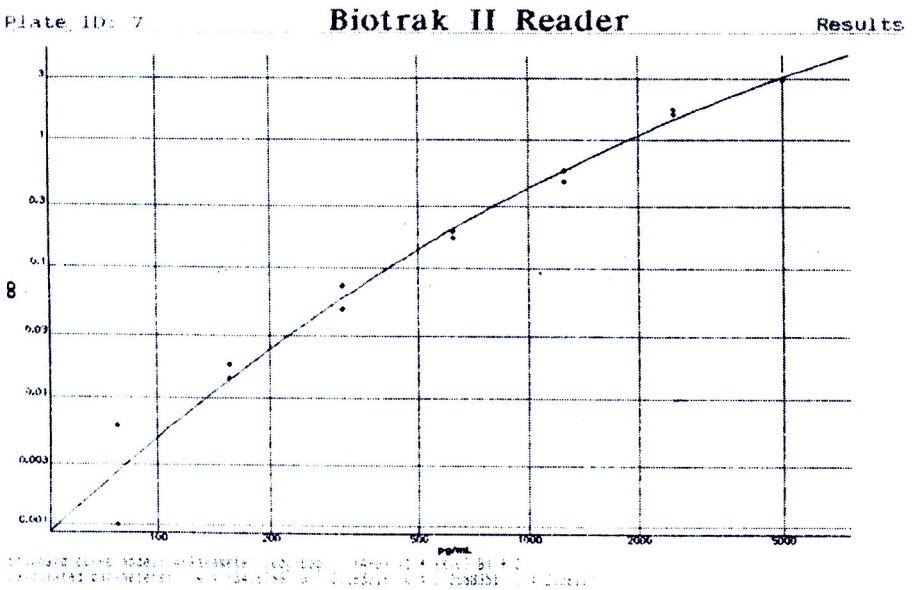

Figure22. Standard curve of DcR3 by ELISA

BIOGRAPHY

Mr. Pramuk Amarinthnukrowh was born in Bangkok, the capital city of Thailand, in November 8th, 1984. In 2007, I received my bachelor degree in Biochemistry from Faculty of Science, Chulalongkorn University. Consequently, with my interests in Human and Molecular Genetics, I had made a decision to study in curriculum of Medical Science in Faculty of Medicine for my Master degree.